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Case Report
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Lipofibromatosis of the Paravertebral Soft Tissue in a Child of Two Years Old

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Abstract

Lipofibromatosis is a bland fibroproliferative process that invades skeletal muscle. Typically the children are affected before 8 years with a peak under 2 years old. Head, neck or thigh are the most frequently involved sites.The management strategies are different: the first, periodical clinical and imaging control with MRI to evaluate the symptoms and the size of the tumor mass; the second, to assess the feasibility and to perform chemotherapy in selected cases; the third, surgical excision of the tumor. This article reports on the youngest patient diagnosed with this rare soft tissue tumor in thoracolumbar area. In fact, according to the Authors' knowledge, no other cases of this tumor in this localization have been presented in the literature. Our study demonstrated that surgical excision is the gold treatment choice in this tumor when feasible. In fact, the scoliosis caused by tumor stopped its evolution after its surgical excision.

Keywords: Lipofibromatosis; Spine; Tumors

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Introduction

There are two types of lipofibromatosis: one type identical to adult desmoidfibromatosis and the second like a diffuse mesenchymal type [1]. Both type often containing fat that usually involves head, neck or thigh. Alternate/Historical Names are: aggressive infantile fibromatosis, cellular fibromatosis, congenital fibromatosis, diffuse mesenchymal fibromatosis, fibrosarcoma-like fibromatosis and infantile desmoid-type fibromatosis[2].

Resonance Imaging (MRI) usually shows a soft tissue lesion with high fat content [3,4].

Excision and histopathological analysis reveal a tumour composed of two patterns [3]: immature or diffuse pattern and uniform bland cells in myxoid stroma (nuclei round or oval to spindled and scant cytoplasm).

There is no evidence for the effectiveness of chemotherapy in this disease and surgical excision seems to be the only possible curative therapy even if recurrences are frequent following incomplete excision [1,5].

The patient here reported was submitted to excisional surgery and resulted in excellent conditions without any sign of recurrence at a follow-up of five years.

Case Report

A.F., male, 29 months old, Caucasian, was referred to our institution with a big painless mass in the thoracolumbar area at right. At the age of 3 months, his parents noticed a "lump" in the right part of the back. At 18.5 month a consultation was performed in another Institution, where a standard radiogram (Figure 1a) showed a thoracolumbar scoliosis (16°Cobb angle with right concavity) and MRI (Figure 2a) described a soft tissue mass in the paraspinal zone from T7 to L4. An ultrasonography-guided biopsy was made at the age of 20 months; histopathologic diagnosis was lipofibromatosis. After this diagnosis the patient underwent chemotherapy with Vinblastine and methotrexate weekly during 7 months. Three gadolinium MRIs were performed to check mass evolution, reporting that the tumour remained unchanged. At the referral to our Institution, the pathologist confirmed to diagnosis of lipofibromatosis. The medical oncologist remarked

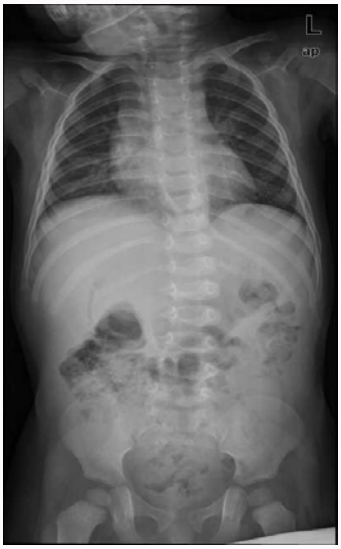

Figure 1a: First X-ray pre op Cobb 16° (18.5 months of life).

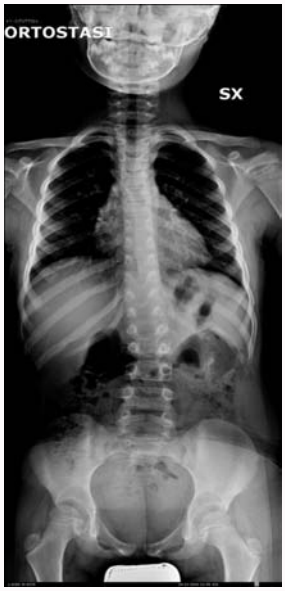

Figure 1b: Latest X-ray performed 5-years after surgery: Cobb angle is conserved and scoliosis was not worsened in comparison with first radiographs.

that further chemotherapy had no supporting evidence. En Bloc resection planned to achieve wide margins was performed at the age of 32 months. The specimen size was 13cm x8cmx5.5 cm. Histology showed an admixture of fibroblastic spindle cells and mature adipose tissue (Figure 3a). Poorly demarcated lobules of fat were separated and traversed by thickened fibroblastic septa. The septa comprised spindle cells showing no atypia or pleomorphism, and minimal amounts of collagen were present (Figure 3b). In addition there were paucicellular fibrotic areas and myxoid change possibly due to chemotherapy. At immunohistochemistry the neoplastic cells were positive for vimentin and negative for smooth muscle act in, desmin, actin muscle specific, caldesmon and beta-catenin. The proliferative index Ki67 (clone MIB1) was 2%.

At five years post-operative follow-up no evidence of local disease was found at clinical controls and also radiological examinations,

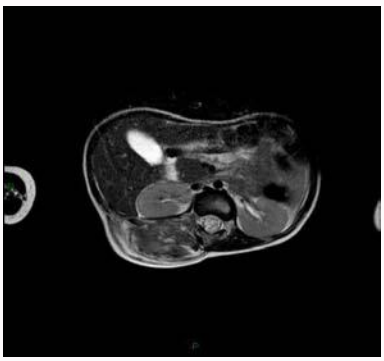

Figure 2a: First MRI preoperative, extension of the lesion in the soft tissues of paravertebral muscles from T7 to L4.

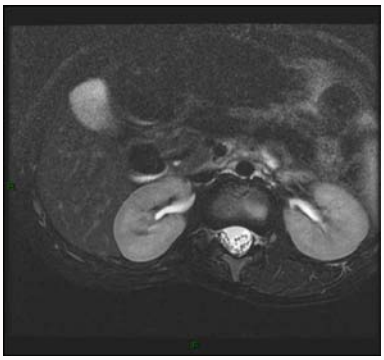

Figure 2b: Post-operative MRI performed 2 years after surgery didn't show local recurrence.

performing during this period, like MRI (Figure 2b) and ultrasound images resulted negative for local recurrence. The scoliosis remained stable and of minor significance even at 5 years of follow-up as demonstrated by the whole spine radiographs performed in orthostatic position at this time that did't show worsening by the first (Figure 1b).

Discussion

Lipofibromatosis is an uncommon tumor typically found on the distal extremities of infants, although it can appear in various sizes and locations andit should be considered in the differential diagnosis of pediatric soft tissue neoplasms [6]. Clinically, the masses are painless and slow-growing [3] but they can cause deformity in the antomical locations where they are present [7]. Surgical excision is the the treatment of choice, while the role of chemotherapy is not supported except in the cases of desmoid-type fibromatosis that is recurrent or not amenable to treatment with surgery [8]. Small number of cases with diagnosis of lipofibromatosis are reported in literature because it is a rare entity.

Fretsch JF et al. [1] described lipofibromatosis for the first time in 2000, as a rare pediatricneoplasm that has been variously interpreted as a type of infantile or juvenile fibromatosis, a variant of fibrous hamartoma of infancy, and a fibrosinglipoblastoma. This variability of interpretations could explain the lack of previous reports. Microscopic examination of 45 cases of soft tissue tumors revealed abundant adipose tissue with a spindled fibroblastic element that chiefly involved the septa of fat and skeletal muscle [9]; these particular findings guide them to propose a new classification for these lesions. In this series regrowth of the tumor or persistent

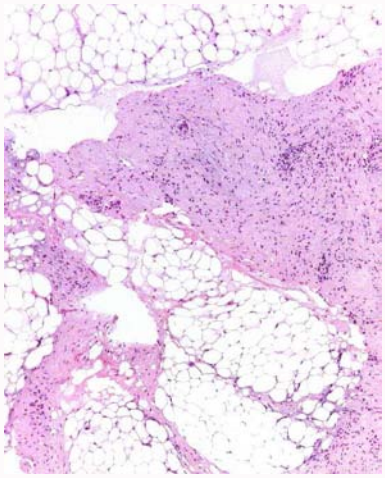

Figure 3a: Histological features: neoplasm constituted of an admixture of fibroblastic spindle cells and mature adipose tissue (magnification 200X).

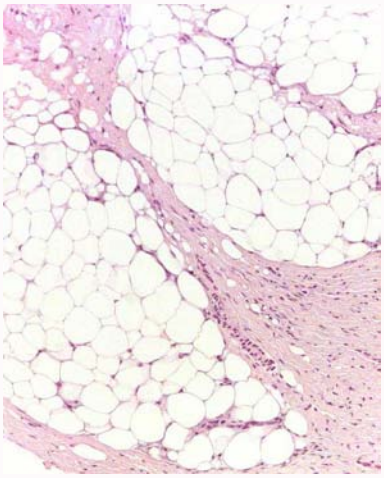

Figure 3b: Histological features: without cytological atypia or pleomorphism (magnification 400X).

disease was documented in 72% of the cases [1]. After this first report, other 6 cases around were registered at present. Marzban and Geramizadeh[10] reported in 2012 a case of lipofibromatosis associated with syndactilia, bilateral complete cleft lip and palate, trigonocephaly, and atrial septal. Herrmann et al. [11] described in 2004 a young girl with a neck mass described as Lipofibromatosis. Friesenbichler and Leithner [12] published in 2010 a case in an adult female of 25 years with a retroclavicular growing lipofibromatosis that caused recurrent paresthesia. Kenney et al. [13] reported in 2007 a case of lipofibromatosis in a 5-year old boy which revealed an apparently balanced three-way translocation, t(4:9:6).

Lipofibromatosis lacks the primitive mesenchymal islands with myxoidstroma that characterize fibrous hamartoma of infancy; discrimination from desmoid-type fibromatosis is based in large part on the degree of preservation of larger fat lobules and the lack of solid fibrous growth. Lipoblastoma can be distinguished based on its constituent of immature adipocytes and the lack of a striking spindle cell component [3,13]. Anomalies involving 6q23~q27 have been implicated in a number of different and seemingly unrelated neoplasms, however, including lymphoid malignancies [14], malignant fibrous histiocytoma[15], angiofibroma [16], and lipomas [17].

This paper reports the youngest patient diagnosed with this rare soft tissue tumor in thoracolumbar area. No other cases with this diagnosis and localization of this tumor are presented in literature. The teaching point of this case is that surgical excision is the gold standard in the treatment of this disease. In fact scoliosis presented at the diagnosis and caused by the presence of lipofibromatosis in the paravertebral soft tissues stopped its evolution after the surgical excision of the tumor and this result was confirmed also at 5 years of follow-up.

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